RNASET2 gene

ribonuclease T2

Normal Function

The *RNASET2* gene provides instructions for making a protein called ribonuclease T2 (RNAse T2), which is abundant in the brain. Ribonucleases help break down RNA, a chemical cousin of DNA. Studies suggest that ribonuclease T2 may also be involved in other functions within cells, such as controlling the development of blood vessels (angiogenesis) and helping to prevent the growth of cancerous tumors. These potential roles of the protein are not well understood.

Health Conditions Related to Genetic Changes

RNAse T2-deficient leukoencephalopathy

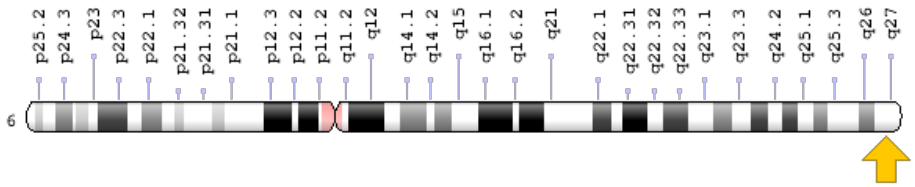
At least 10 *RNASET2* gene mutations have been identified in people with RNAse T2-deficient leukoencephalopathy. This disorder involves brain abnormalities leading to neurological problems that become apparent during infancy, affecting intellectual ability and the development of motor skills such as sitting and crawling.

The *RNASET2* gene mutations that cause RNAse T2-deficient leukoencephalopathy result in loss of ribonuclease T2 protein function. It is unknown how loss of this protein results in the brain abnormalities and neurological problems characteristic of RNAse T2-deficient leukoencephalopathy. Researchers have noted that the signs and symptoms of RNAse T2-deficient leukoencephalopathy are similar to those resulting from infection by a particular virus, called cytomegalovirus (CMV), when it is transmitted to a fetus during pregnancy (congenital CMV). They are seeking to understand how the viral infection, or the body's response to it, and the loss of ribonuclease T2 function could have similar effects on the developing brain. It is thought that both may be related to changes in angiogenesis or an immune system response to RNA that has not been properly broken down.

Chromosomal Location

Cytogenetic Location: 6q27, which is the long (q) arm of chromosome 6 at position 27

Molecular Location: base pairs 166,929,509 to 166,956,589 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- bA514O12.3
- FLJ10907
- ribonuclease 6
- ribonuclease T2 precursor
- RNASE6PL

Additional Information & Resources

Educational Resources

- Angiogenesis (2010): Overview of Angiogenesis
  https://www.ncbi.nlm.nih.gov/books/NBK53238/
- The Cell: A Molecular Approach (second edition, 2000): Tumor Suppressor Genes
  https://www.ncbi.nlm.nih.gov/books/NBK9894/

GeneReviews

- Leukodystrophy Overview
  https://www.ncbi.nlm.nih.gov/books/NBK184570

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28RNASET2%5BTIAB%5D%29+OR+%28RNASE6PL%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

OMIM

- RIBONUCLEASE T2
  http://omim.org/entry/612944

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/RNASET2ID518ch6q27.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=RNASET2%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=21686
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/8635
- UniProt
  http://www.uniprot.org/uniprot/O00584

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